Improvement of Kearns-Sayre Syndrome with Controlled Carbohydrate Intake and Coenzyme Q₁₀ Therapy

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Dear Sir,

We read the interesting paper of Tsuyama et al. [1] and would like to add our therapeutic experience with a case of Kearns-Sayre syndrome with hyperlactacidemia.

A boy, 9 years and 9 months old, complained of bilateral ptosis, ophthalmoplegia, gait disturbance, atypical chorioretinal degeneration, muscle weakness and hyporeflex-ia (fig. 1, 2). Blood lactate, pyruvate and serum creatinine phosphokinase were 30 mg/dl (normal 6-16 mg/dl), 0.22 mg/dl (normal 0.3-0.5 mg/dl) and 274 IU/dl (normal up to ! -0 IU/dl), respectively. A muscle biopsy (nui > -culus quadriceps femoris) showed neither ragged red fibers nor necrosis of fibers. EMU was of the myopathic type. On the basis of the criteria of Rowland et al. [2], a diagnosis of Keams-Sayre syndrome was made. We assumed an overload of metabolites (lactate) preceding the defect in energy metabolism and a chronic deficiency of end products (coenzyme Q), as a pathogenetic mechanism.

A diet with carbohydrate intake of 7.5 g/ kg/day (minimal dietary allowance), with fat intake of 3.2 g/kg/day (high fat) was administered with the purpose of lowering the high lactate levels. After 3 months, the fasting serum lactate became normal and the ophthalmoplegia, the choreiform movements, the ataxia and the psychomotor disturbances improved.

Thereafter, with an unchanged diet, coenzyme Q₁₀ ('Juvacor', Invemì della Beffa, under licence of Taiyo Pharmaceutical Industry, Tokyo, Japan) therapy was started (30 mg daily) [3]. After 3 months, the fasting lactate was

Fig.1. 2-year-old patient.

6.6 mg/dl, the choreiform movements disappeared, bilateral ptosis improved and eye movements were present (fig. 3a-c) with only a mild ophthalmoparesis. This condition persists after 6 months of unmodified diet and therapy. In another personal case of Kearns-

Fig. 2. Patient aged 9 years and 9 months.

Sayre syndrome, no clinical improvement was observed after therapy with coenzyme Q alone.

We think that in some cases of Kearns-Sayre syndrome an association of a diet with low-normal carbohydrate, high fat intake and coenzyme Q₁₀ therapy may be useful.

References

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